What is claimed is:

1. Substantially pure nucleic acid encoding a mammalian methionine synthase reductase polypeptide.

2. The nucleic acid of claim 1, wherein said nucleic acid encodes a human polypeptide.

3. The nucleic acid of claim 1, wherein said nucleic acid has the sequence of SEQ ID NO: 1 or SEQ ID NO: 41, or degenerate variants thereof, and wherein said nucleic acid encodes the amino acid sequence of SEQ ID NO: 2 or SEQ ID NO: 42.

4. A substantially pure nucleic acid that hybridizes at high stringency to a sequence found within the nucleic acid of SEQ ID NO: 1 or SEQ ID NO: 41.

5. The nucleic acid of claim 4, wherein said nucleic acid has a sequence complementary to at least 50% of at least 60 nucleotides of the nucleic acid encoding the methionine synthase reductase polypeptide, said sequence sufficient to allow nucleic acid hybridization under high stringency conditions.

6. The nucleic acid of claim 5, wherein said nucleic acid contains a mutation or a polymorphism, and wherein said nucleic acid encodes a mutant or polymorphic polypeptide or a fragment thereof.

7. The mutation of claim 6, wherein said mutation is a 4 base deletion starting from base 1675 of SEQ ID NO: 1.

8. The mutation of claim 6, wherein said mutation is a deletion of 3 bases starting from base 1726 of SEQ ID NO: 1.

9. A non-human animal wherein one or both genetic alleles encoding a methionine synthase reductase polypeptide are mutated.

10. The animal of claim 9, wherein one or both genetic alleles encodes a methionine synthase reductase polypeptide are disrupted, deleted, or otherwise rendered nonfunctional.

11. The animal of claim 9, wherein said polypeptide has a mutation associated with hyperhomocysteinemia.

12. The animal of claim 9, wherein said animal is a rodent or a nematode.

13. An antibody that specifically binds a methionine synthase reductase polypeptide.

14. A method of detecting the presence of a methionine synthase reductase polypeptide, said method comprising contacting a sample with the antibody that specifically binds a methionine synthase reductase polypeptide and assaying for binding of said antibody to said polypeptide.

15. A method for detecting sequence variants for methionine synthase

reductase in a mammal, said method comprising analyzing the nucleic acid of a test subject to determine whether said test subject contains a mutation or polymorphism in a methionine synthase reductase gene, wherein the presence of said mutation or said polymorphism is an indication that said animal has an increased or decreased likelihood of developing hyperhomocysteinemia, cardiovascular disease, neural tube defects, or cancer.

16. A method of treating or preventing cancer, cardiovascular disease, or neural tube defects in a subject, said method comprising inhibiting methionine synthase reductase biological activity in said subject.

17. A method of treating or preventing cardiovascular disease, said method comprising administering to the subject to a therapeutically effective dose of a metabolite or cofactor selected from the group: folate, cobalamin, S-adenosyl methionine, betaine, or methionine.

18. The method of claim 16 or 17, wherein said subject has been diagnosed as having a mutation or polymorphism in methionine synthase reductase.

19. A method of preventing neural tube defects, cancer, or cardiovascular disease, said method comprising:

a) detecting an increased risk of neural tube defects, cancer, or cardiovascular disease, wherein said detecting is by analyzing methionine synthase reductase nucleic acid from one or more test subjects selected from: a mammal; a

potential parent, either male or female; a pregnant mammal; or a developing embryo or fetus, wherein said analyzing is done by the method of claim D; and

b) exposing said mammal, said potential parent, said pregnant mammal, and/or said developing embryo or fetus to a therapeutically effective dose of a metabolite or cofactor selected from the group: cobalamin; S-adenosyl methionine; betaine; or methionine, wherein said exposing is via the administration of said dose to said mammal, said potential parent, said pregnant mammal, and/or said developing embryo or fetus.

20. A method for screening for a compound that modulates methionine synthase reductase biological activity, said method comprising the steps of:

a) contacting a sample containing mutated or polymorphic methionine synthase reductase with said compound, and

b) assaying for methionine synthase reductase enzymatic activity, wherein increased enzymatic activity indicates an inducer of methionine synthase reductase biological activity, and decreased enzymatic activity indicates an inhibitor of methionine synthase reductase biological activity.

21. A method for screening for a compound that modulates methionine synthase reductase biological activity, said method comprising the steps of:

a) contacting a sample with said compound, and

b) assaying for methionine synthase reductase expression, wherein increased expression indicates an inducer of methionine synthase reductase biological activity, and decreased expression indicates an inhibitor of methionine synthase reductase biological activity.

22. A method for detecting an increased risk of developing a neural tube defect in a mammalian embryo or fetus, said method comprising detecting the presence of a polymorphic methionine synthase reductase (MTRR) in a test subject, wherein said polymorphic MTRR contains a methionine instead of an isoleucine at amino acid position 22, wherein said test subject is a future parent of said embryo or said fetus, and wherein detection of a homozygous MTRR polymorphism in said future parent, said embryo, or said fetus, or detection of either a homozygous or heterozygous MTRR polymorphism in both future parents, indicates an increased risk of developing said neural tube defect in said embryo or said fetus.

23. The method of claim 22, wherein said polymorphic MTRR is detected by analyzing nucleic acid from said test subject.

24. The method of claim 23, wherein said nucleic acid is genomic DNA.

25. The method of claim 23, wherein said nucleic acid is cDNA.

26. The method of claim 23, wherein said nucleic acid contains a G instead of an A at the third position of the twenty-second codon (nucleotide position 66, relative to the first nucleotide of the start codon) of MTRR.

27. The method of claim 23, said method further comprising:

a) PCR-amplifying a segment of MTRR nucleic acid using

primers MSG108S (SEQ ID NO: 49) and AD292 (SEQ ID NO: 50), and

b) digesting the product of the PCR amplification reaction with the restriction enzyme *Nde* I, wherein a PCR product that is digested by *Nde* I indicates an increased risk of developing a neural tube defect in a mammalian embryo or fetus.

28. The method of claim 22, wherein said polymorphic MTRR is detected by analyzing MTRR polypeptide from said test subject.

29. The method of claim 22, wherein said test subject is a future female parent of said embryo or said fetus.

30. The method of claim 22, wherein said test subject is said embryo or said fetus.

31. The method of claim 22, said method further comprising detecting the presence of a polymorphic methylenetetrahydrofolate reductase (MTHFR) in a test subject, said polymorphic MTHFR having a T instead of a C at a nucleotide position equivalent to position 677 of SEQ ID NO: 51, wherein detection of said polymorphic MTHFR indicates an increased risk of developing said neural tube defect in said embryo or said fetus.

32. The method of claim 31, wherein said polymorphic MTHFR is detected by analyzing nucleic acid from said test subject.

33. The method of claim 31, wherein said polymorphic MTHFR is detected by analyzing polypeptide from said test subject.

34. The method of claim 22, said method further comprising measuring the level of cobalamin in said test subject, wherein a low cobalamin level indicates an increased risk of developing said neural tube defect in said embryo or said fetus.